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DIAGNOSTIC METHOD FOR POMPE'S DISEASE

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(57) Claim

1. A method of diagnosis of Pompe's disease in cattle and in particular Brahman based cattle including the steps of:-

- (i) obtaining a purified sample of test DNA from a subject suspected of carrying or suffering from Pompe's disease;
- (ii) mixing the purified test DNA with a restriction enzyme recognising the nucleotide sequence 5'...CCGG...3';
- (iii) separating DNA fragments obtained after step (ii); and
- (iv) hybridising the fragments with a probe based on the GAA gene nucleotide sequence.

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10. A method of diagnosis of Pompe's Disease in cattle and in particular Brahman based cattle including the steps of:-

- (i) obtaining a sample of DNA from cattle suspected of having Pompe's disease;
- (ii) mixing the sample with PCR primers derived from bovine GAA sequence data which effectively amplifies DNA across a RFLP site associated with Pompe's disease;
- (iii) reacting the mixture obtained in step (ii) in the presence of a suitable buffer, dATP, dCTP, dGTP and dTTP in combination with a suitable polymerase;
- (iv) cutting the amplified PCR reaction mix with a restriction enzyme recognising the nucleotide sequence 5'...CCGG...3'; and
- (v) analysing the reaction mixture after step (iv) to ascertain the presence of Pompe's disease genotypes.

11. A method as claimed in claim 10, wherein the restriction enzyme is selected from the group Bsu 192 I, Bau 123 I, Hap II, Hpa II, Mni II, MnoI, MspI and Sta GUI.